NIH Eunice Kennedy Shriver National Institute of Child Health and Human Development
Healthy pregnancies. Healthy children. Healthy and optimal lives.

Turner Syndrome

Turner syndrome is a condition in which a girl or woman is partially or completely missing an X chromosome. It can cause infertility and heart problems and alter a female's appearance. NICHD plays a leading role in advancing research on Turner syndrome by supporting the investigation of its physical and emotional effects as well as potential therapies.

About Turner Syndrome

Turner syndrome is a disorder caused by a partially or completely missing X chromosome. This condition affects only females.

Most people have 46 chromosomes in each cell—23 from their mother and 23 from their father. The 23rd pair of chromosomes are called the sex chromosomes—X and Y—because they determine whether a person is male or female. Females have two X chromosomes (XX) in most of their cells, and males have one X chromosome and one Y chromosome (XY) in most of their cells. A female with all of her chromosomes is referred to as 46,XX. A male is 46,XY.

Turner syndrome most often occurs when a female has one normal X chromosome, but the other X chromosome is missing (45,X). Other forms of Turner syndrome result when one of the two chromosomes is partially missing or altered in some way.[1]

Citations

1. Turner Syndrome Society. (n.d.). About Turner Syndrome. https://www.turnersyndrome.org/about-turnersyndrome

What are the symptoms of Turner syndrome?

Turner syndrome causes a variety of symptoms in girls and women. For some people, symptoms are mild, but for others, Turner syndrome can cause serious health problems. In general, women with Turner syndrome have female sex characteristics, but these characteristics are underdeveloped compared to the typical female. Turner syndrome can affect:[1]

- **Appearance.** Features of Turner syndrome may include a short neck with a webbed appearance, low hairline at the back of the neck, low-set ears, hands and feet that are swollen or puffy at birth, and soft nails that turn upward.
- **Stature.** Girls with Turner syndrome grow more slowly than other children. Without treatment, they tend to have short stature (around 4 feet, 8 inches) as adults.
- **Puberty.** Most girls with Turner syndrome do not start puberty naturally.
- **Reproduction.** In most girls with Turner syndrome, the ovaries are missing or do not function properly. Without the estrogen made by their ovaries, girls with Turner syndrome will not develop breasts. Most women with Turner syndrome cannot become pregnant without assistive technology.[2]
- **Cardiovascular.** Turner syndrome can cause problems with the heart or major blood vessels. In addition, some women and girls with Turner syndrome have high blood pressure.
- **Kidney.** Kidney function is usually normal in Turner syndrome, but some people with this condition have kidneys that look abnormal.
- **Osteoporosis.** Women with Turner syndrome often have low levels of the hormone estrogen, which can put them at risk for osteoporosis. Osteoporosis can cause height loss and bone fractures.
- **Diabetes.** People with Turner syndrome are at higher risk for type 2 diabetes.
- **Thyroid.** Many people with Turner syndrome have thyroid issues. The most common one is hypothyroidism, or an underactive thyroid gland.
- **Cognitive.** People with Turner syndrome have normal intelligence. Some, however, have challenges learning mathematics or with visual-spatial coordination (such as determining the relative positions of objects in space).

Citations

1. Turner Syndrome Society. (2017). Clinical practice guidelines for the care of women and girls with Turner syndrome. European Society of Endocrinology, 117:3, G1-G70. Retrieved 11/29/2017 from http://docs.wixstatic.com/ugd/8fb9de_905ef4f4146a487a9f7031a319b85fe2.pdf (PDF 1.4 MB).
2. Intersex Society of North America. (n.d.). *Turner syndrome*. Retrieved June 14, 2012, from http://www.isna.org/faq/conditions/turner
3. Bondy, C. A. (2007). Care of girls and women with Turner syndrome: A guideline of the Turner Syndrome Study Group. *Journal of Clinical Endocrinology & Metabolism, 92*, 10-25.

How many people are affected or at risk of Turner syndrome?

Turner syndrome affects about 1 of every 2,500 female live births worldwide.[1]

This disorder affects all races and regions of the world equally. There are no known environmental risks for Turner syndrome. Parents who have had many unaffected children can still have a child with Turner syndrome later on.

Generally, Turner syndrome is not passed on from mother to child. In most cases, women with Turner syndrome are infertile.

Citations

1. Turner Syndrome Society. (n.d.). *What is Turner syndrome? Fact sheet*. Retrieved July 16, 2012, from https://www.turnersyndrome.org/about-turnersyndrome

What causes Turner syndrome?

Turner syndrome occurs when part or all of an X chromosome is missing from most or all of the cells in a girl's body. A girl normally receives one X chromosome from each parent. The error that leads to the missing chromosome appears to happen during the formation of the egg or sperm.

Most commonly, a girl with Turner syndrome has only one X chromosome. Occasionally, she may have a partial second X chromosome. Because she is missing part or all of a chromosome, certain genes are missing. The loss of these genes leads to the symptoms of Turner syndrome.[1]

Sometimes, girls with Turner syndrome have some cells that are missing one X chromosome (45,X) and some that are normal. This is because not every cell in the body is exactly the same, so some cells might have the chromosome, while others might not. This condition is called mosaicism (pronounced *moh-ZEY-uh-siz-uhm*). If the second sex chromosome is lost from most of a girl's cells, then it's likely that she will have symptoms of Turner syndrome. If the chromosome is missing from only some of her cells, she may have no symptoms or only mild symptoms.

Citations

1. National Human Genome Research Institute. (2011). *Learning about Turner syndrome*. Retrieved June 14, 2012, from https://www.genome.gov/Genetic-Disorders/Turner-Syndrome

How do healthcare providers diagnose Turner syndrome?

Healthcare providers use a combination of physical symptoms and the results of a genetic blood test, called a karyotype, to determine the chromosomal characteristics of the cells in a female's body. The test will show if one of the X chromosomes is partially or completely missing.

Turner syndrome also can be diagnosed during pregnancy by testing the cells in the amniotic fluid. Newborns may be diagnosed after heart problems are detected or after certain physical features, such as swollen hands and feet or webbed skin on the neck, are noticed. Other characteristics, like widely spaced nipples or low-set ears, also may lead to a suspicion of Turner syndrome. Some girls may be diagnosed as teenagers because of a slow growth rate or a lack of puberty-related changes. Still others may be diagnosed as adults when they have difficulty becoming pregnant.[1]

Citations

1. National Human Genome Research Institute. (2011). *Learning about Turner syndrome*. Retrieved July 14, 2012, from https://www.genome.gov/Genetic-Disorders/Turner-Syndrome

What are common treatments for Turner syndrome?

Although there is no cure for Turner syndrome, some treatments can help minimize its symptoms. These include[1]:

- **Human growth hormone.** If given in early childhood, hormone injections can often increase adult height by a few inches.
- **Estrogen replacement therapy (ERT).** ERT can help start the secondary sexual development that normally begins at puberty (around age 12). This includes breast development and the development of wider hips. Healthcare providers may prescribe a combination of estrogen and progesterone to girls who haven't started menstruating by age 15. ERT also provides protection against bone loss.

Regular health checks and access to a wide variety of specialists are important to care for the various health problems that can result from Turner syndrome.[2] These include ear infections, high blood pressure, and thyroid problems.

Citations

1. National Human Genome Research Institute. (2011). *Learning about Turner syndrome*. Retrieved July 17, 2012, from https://www.genome.gov/Genetic-Disorders/Turner-Syndrome
2. Bondy, C. A. (2007). Care of girls and women with Turner syndrome: A guideline of the Turner Syndrome Study Group. *Journal of Clinical Endocrinology & Metabolism, 92*, 10-25.

Turner Syndrome | NICHD - Eunice Kennedy Shriver National Institute of Child Health and Human Development
NICHD Information Resource Center
Phone: 1-800-370-2943
Email: NICHDInformationResourceCenter@mail.nih.gov
Fax: 1-866-760-5947
Mail: P.O. Box 3006, Rockville, MD 20847
For the Federal Relay Service, dial 7-1-1